The Chado “Health” module

FlyBase

FB2013_06, released November 1st, 2013

A Database of *Drosophila* Genes & Genomes

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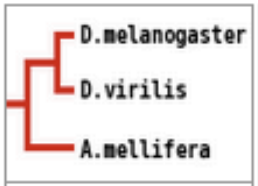
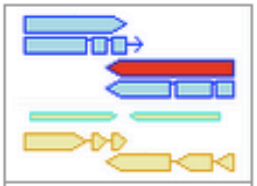
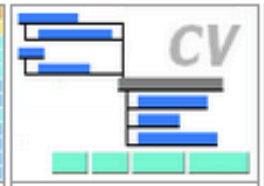


BLAST


GBrowse

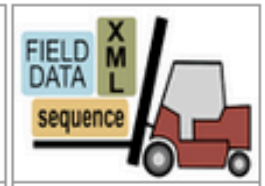
QueryBuilder

RNA-Seq Search



TermLink


ImageBrowse



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FlyBase Forum

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News

FlyBase 102 | 1 Dec 13
New in Release FB2013_06 | 4 Nov 13
2014 Dros Image Award | 22 Oct 13
Stock Center User Survey | 18 Jun 13
Fly Mating Scheme Design | 13 Mar 13
GenBank Release | 25 Jan 13
2013 Release Schedule | 6 Nov 12
NAR article on bibliography | 5 Nov 12
White Paper 2012 | 8 Jun 12

Upcoming Meetings

Invtbt-Microbe Interactions | 26 Jan 14
Joint SFBD-EFOR meeting | 10 Feb 14
55th ADRC | 26 Mar 14
19th Crete Dros Mol/Dev Bio | 22 Jun 14
7th Intl Symp Mol Insect Sci | 13 Jul 14
Dros in Exp Genetics & Bio | 6 Oct 14
2nd FSE-EMBO Minibrains | 8 Nov 14

QuickSearch

Simple | Expression | Phenotype | GO | References | Data Class

°Author(s) : e.g. 'Smith NOT Johnson'

Year(s) : e.g. '2004-2008', '>2001', etc.

Title/Abstract : search Title and Abstract text

°Journal : e.g. 'Dev. Biol.'

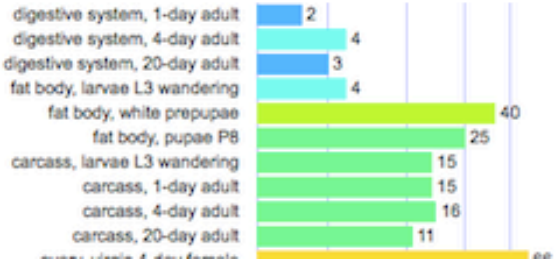
Search

°QuickSearch autocomplete: ☐ **Note:** Wild cards (*) can be added to your search term

Commentary

See all commentaries

FlyBase RNA-Seq RPKM data bulk download



May 6, 2013. FlyBase is extending its initial gene-level analyses of RNA-seq throughput data from modENCODE and others. The algorithm for RPKM (reads per kilobase per million mapped reads) has been refined, additional datasets have been analyzed, and these data are now available for bulk download... (More)

FlyBase use case

Drosophila used to model aspects of a human disease or disorder

Provide links to information about the condition - Human Disease Summary

Support searches via synonyms, dbxrefs and cvterms

Link to reagents – fly alleles and transgenic constructs (human genes in fly)

Link to literature

Link to fly phenotypes

General Information			
Name HH1b	amyotrophic lateral sclerosis 10	FlyBase ID HH1f	FBds0000005
DOID HH2d		Parent Disease HH2b (HH1b of)	amyotrophic lateral sclerosis
OMIM HH1c/HH2c	ALS10 612069	Parent Disease DOID HH2b (HH2d of)	DOID:332
Disease Summary Information			
Parent Disease Summary: amyotrophic lateral sclerosis HH2b (HH1b of)			
Symptoms and phenotype HH2b (HH4a of)	Amyotrophic lateral sclerosis is a neurodegenerative disorder characterized by the death of motor neurons in the brain, brainstem, and spinal cord, resulting in fatal paralysis. ALS usually begins with asymmetric involvement of the muscles in middle adult life. Approximately 10% of ALS cases are familial. ALS is sometimes referred to as 'Lou Gehrig disease' after the famous American baseball player who was diagnosed with the disorder. [from OMIM 105400, 2013.08.02]		
Genetics HH2b (HH4b of)	ALS is a genetically heterogeneous disorder, with several causative genes and mapped loci. [from OMIM 105400, 2013.08.02]		
Specific Disease Summary: amyotrophic lateral sclerosis 10 HH1b			
Human gene(s) implicated HH7a	Hsap\TARDBP		
Symptoms and phenotype HH4a	Some patients with mutations in the TARDBP gene develop frontotemporal dementia with TDP43-positive inclusions. Patients with TARDBP mutations and frontotemporal dementia may or may not have associated signs of motor neuron disease. [from OMIM 612069, 2013.08.02]		
Genetics HH4b	This form of autosomal dominant amyotrophic lateral sclerosis, ALS10, is caused by heterozygous mutation in the TARDBP gene, which encodes the TDP43 protein, on chromosome 1p36. [from OMIM 612069, 2013.08.02]		

Model Status and Key Findings

Status of Drosophila Model

HH10	Dmel\@TBPH@ is similar in structure and in vitro functions to the human gene TARDBP. Dmel\@TBPH@ mutants exhibit adult locomotor defects. Tissue-specific expression of transgenic constructs of @Hsap\TARDBP@ results in a neurodegeneration phenotype, reduced life span, and/or locomotor defects. Cytoplasmic, but not nuclear, accumulation of @Hsap\TARDBP@ is cytotoxic. @Hsap\TARDBP@ interacts genetically with @Hsap\UBQLN1@. Dmel\@TBPH@ interacts physically with Dmel\@caz@. [updated August 2013]

Description of experiments

Mammalian transgenic(s) HH11a	Drosophila transgenic lines of a mammalian orthologous gene have been created (FBrf0210043, FBrf0210443, FBrf0210957, FBrf0211667, FBrf0212655, FBrf0213887, FBrf0214156, FBrf0214718, FBrf0216240, FBrf0217614, FBrf0219102).

Useful Dmel transgene(s) HH15b

Transgene	Allele	Associated Stock
P{UAS-TBPH.L}	TBPH$^{Scer\UAS.cLa}$	Not publicly available
P{UAS-TBPH.R}	TBPH$^{Scer\UAS.cRa}$	Not publicly available

Stocks from Dmel RNAi collections HH15d

Transgene	Allele	Associated Stock
P{TRiP.HM05194}	TBPH^{HM05194}	P{TRiP.HM05194}attP2
P{GD6943}	TBPH^{GD6943}	P{GD6943}v38377 P{GD6943}v38379
P{GD17843}	CG7804^{GD17843}	P{GD17843}v49886 P{GD17843}v49887

Useful Classical Alleles of Drosophila Orthologs HH15c

Allele	Allele Class	Mutagen	Associated Stock(s)
TBPH^{ex26}	Amorphic allele	P-element activity	Not publicly available
TBPH^{EY10530}		P-element activity	P{EPgy2}TBPH^{EY10530}
TBPH^{G2}	Amorphic allele	P-element activity	Not publicly available

References

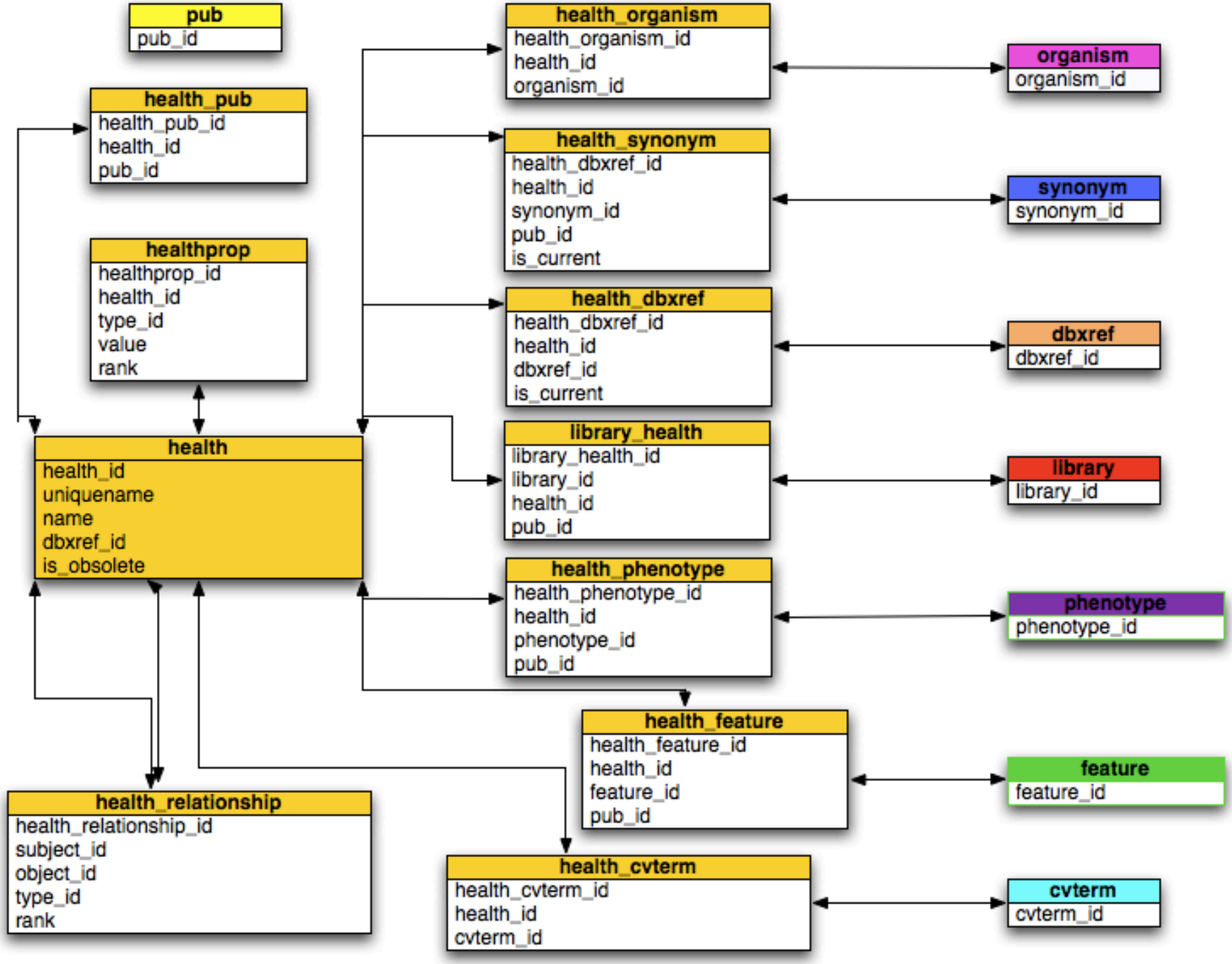
Research Papers	Gregory et al., 2012, PLoS ONE 72): e31899 The Aggregation and Neurotoxicity of TDP-43 and Its ALS-Associated

Other use cases

Crop pathology

Animal disease

???


pub
pub_id
health_pub
health_pub_id
health_id
pub_id
healthprop
healthprop_id
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type_id
value
rank
health
health_id
uniquename
name
dbxref_id
is_obsolete
health_relationship
health_relationship_id
subject_id
object_id
type_id
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health_organism
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Phenotype ?

- We do not want to capture detail about the human disease phenotypes per se.
- We do want to be able to link observable fly phenotypes with the human disease – i.e. relating to disease model
- To support searches we would need to add non-optimal linking tables eg. phenotype_synonym

```
-- ============================================
-- TABLE: phenotype
-- ============================================

CREATE TABLE phenotype (
    phenotype_id SERIAL NOT NULL,
    primary key (phenotype_id),
    uniquename TEXT NOT NULL,
    name TEXT default null,
    observable_id INT,
    FOREIGN KEY (observable_id) REFERENCES cvterm (cvterm_id) ON DELETE CASCADE,
    attr_id INT,
    FOREIGN KEY (attr_id) REFERENCES cvterm (cvterm_id) ON DELETE SET NULL,
    value TEXT,
    cvalue_id INT,
    FOREIGN KEY (cvalue_id) REFERENCES cvterm (cvterm_id) ON DELETE SET NULL,
    assay_id INT,
    FOREIGN KEY (assay_id) REFERENCES cvterm (cvterm_id) ON DELETE SET NULL,
    CONSTRAINT phenotype_c1 UNIQUE (uniquename)
);
CREATE INDEX phenotype_idx1 ON phenotype (cvalue_id);
CREATE INDEX phenotype_idx2 ON phenotype (observable_id);
CREATE INDEX phenotype_idx3 ON phenotype (attr_id);

COMMENT ON TABLE phenotype IS 'A phenotypic statement, or a single
atomic phenotypic observation, is a controlled sentence describing
observable effects of non-wild type function. E.g. Obs=eye, attribute=color, cvalue=red.';
COMMENT ON COLUMN phenotype.observable_id IS 'The entity: e.g. anatomy_part, biological_process.';
COMMENT ON COLUMN phenotype.attr_id IS 'Phenotypic attribute (quality, property, attribute, character) - drawn from PATO.';
COMMENT ON COLUMN phenotype.value IS 'Value of attribute - unconstrained free text. Used only if cvalue_id is not appropriate.';
COMMENT ON COLUMN phenotype.cvalue_id IS 'Phenotype attribute value (state).';
COMMENT ON COLUMN phenotype.assay_id IS 'Evidence type.';
```

-- TABLE: phenotype_cvterm

-- TABLE: feature_phenotype

-- TABLE: phenotypeprop

\+ Genetic module

FlyBase

FB2013_06, released November 1st, 2013

Precomputed data files for the current release

Help Open All Close All

Main Data Set

Postgres Chado Database Dump

Chado database (ftp://ftp.flybase.net/releases/FB2013_06/psql)

Drosophila Data

Current FTP repository (ftp://ftp.flybase.net/releases/FB2013_06/)

Current Chado-XML repository (ftp://ftp.flybase.net/releases/FB2013_06/chado-xml)

Current Reporting-XML repository (ftp://ftp.flybase.net/releases/FB2013_06/reporting-xml)

Genomes FTP archive (ftp://ftp.flybase.net/genomes/)

- **Synonyms**
- **Genes**
- **Alleles and Stocks**
- **Nomenclature**
- **Ontology Terms**
- **Genomes: Annotation and Sequence**
- **Transcripts and Polypeptides**
- **Transposons, Transgenic Constructs, and Insertions**
- **Aberrations**
- **Large Dataset Metadata**
- **Clones**
- **References**
- **Drosophila Researchers**
- **Map Conversion Tables**